PP-020

Primary hyperoxaluria in an adult presenting with end stage renal failure, without history of nephrocalcinosis or nephrolithiasis

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Introduction: Primary hyperoxaluria (PH) is a rare genetic disorder characterized by overproduction of oxalate due to specific enzyme deficiencies in glyoxylate metabolism. The primary clinical presentation is in the form of recurrent urolithiasis, progressive nephrocalcinosis, end-stage renal disease, and systemic oxalosis.

Case report: We report 48 years-old women with a family history of PH, were admitted to the hospital for vomiting, abdominal pain and anorexia. Physical examination showed hypertension (blood pressure=15/10 cmH2O), the rest of examination was normal. Laboratory data, revealed kidney failure; serum creatinine=1970µmol/l, hemoglobin=7g/dl, Calcium=1.37mmol/l, Phosphore=2.32mmol/l, PTH (parathormone)= 416.4pg/l

24h proteinuria was negative. Crystalluria and infrared spectroscopy showed CaOx monohydrate crystals (type Ic whewellite). Renal ultrasound didn't show nephrocalcinosis or nephrolithiasis. Genetic analysis of AGXT gene allow us to detect homozygote mutations in the allele AGXT33-34 ins C.

End stage of renal disease was admitted and the patient required Hemodialysis. Exploration of other organs didn't show abnormalities.

Discussion and conclusion: Here we report an exceptional case of oxalosis diagnosed at a later age and without a history of renal calculi or extra renal manifestations.

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